TRANSMISSION GENETICS

Single-Gene Inheritance

Gregor Mendel

- Father of genetics
- Demonstrated that the inheritance of certain traits in pea plants follows particular patterns
  - Why pea plants?
    - Easy to grow
    - Develop quickly
    - Have many traits that take 1 of 2 easily distinguishable forms

Mendel Studied Transmission of Seven Traits in the Pea Plant

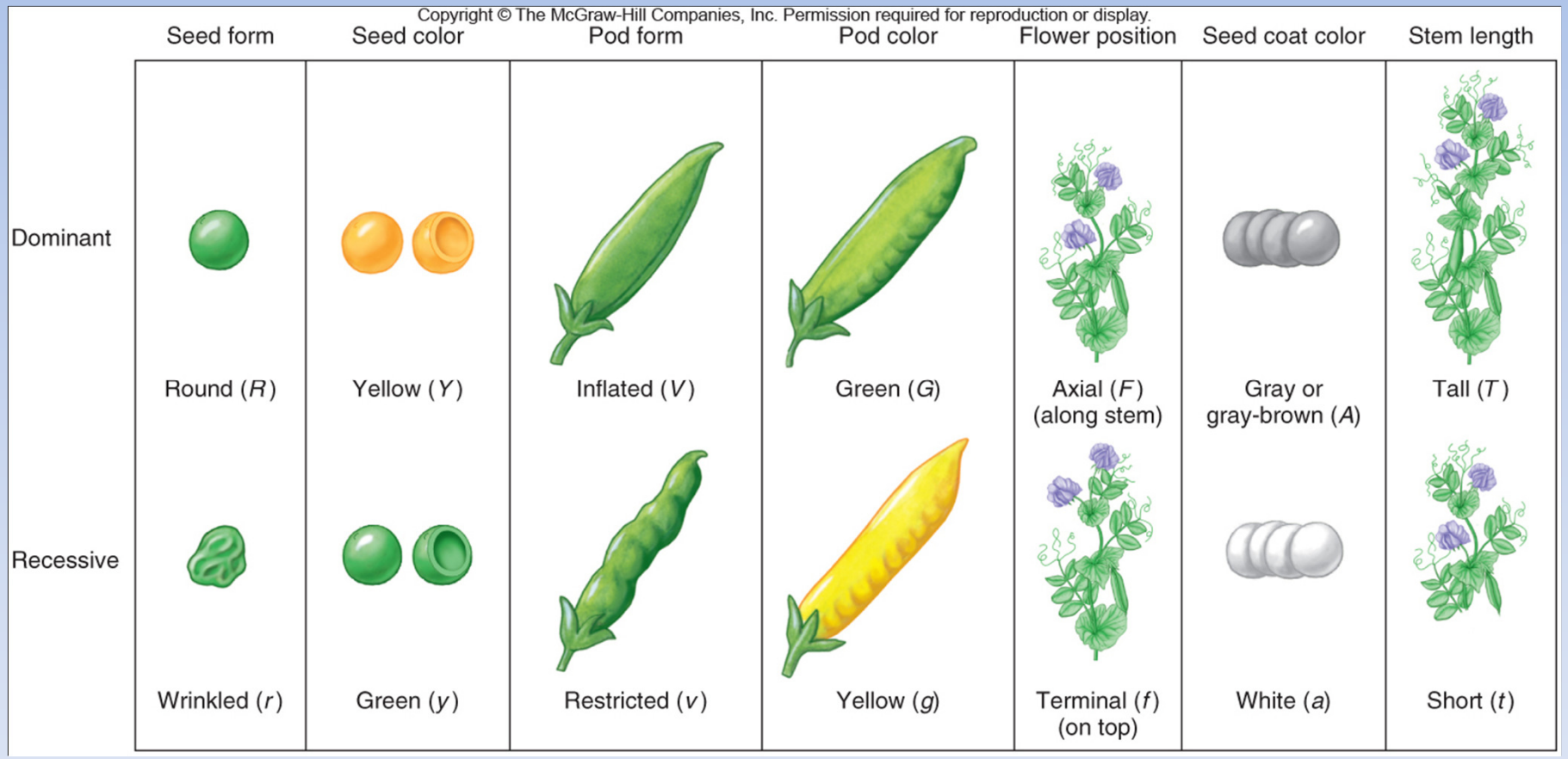


Figure 4.2

Figure 4.1

Vocabulary

- ***Gene*** =
  - Unit by which hereditary characteristics are transmitted

- ***Allele*** =
  - An alternate (variant) form of a gene

- ***Dominant*** =
  - An allele expressed when present in one copy
  - Represented with a capital letter

- ***Recessive*** =
  - An allele whose expression is masked by another allele
  - Represented with a lowercase letter

- ***Homozygous*** =
  - Having 2 identical alleles of a gene
    - Either both dominant or both recessive
  - AKA “true-breeding”
  - Examples
    - TT
    - tt

- ***Heterozygous*** =
  - Having 2 different alleles of a gene
    - One dominant and one recessive
  - AKA “non-true-breeding” or “hybrid”
  - Example
    - Tt

- ***Genotype*** =
  - The allele combinations in an individual that cause particular traits or disorders
- ***Phenotype*** =
  - The expression of a gene in traits or symptoms
- ***Wild type*** =
  - The most common phenotype in a population for a particular gene
- ***Mutant*** =
  - An allele that differs from the normal or most common allele in a population that alters the phenotype

Practice Examples:
Writing Genotypes and Phenotypes

Tall (T) is dominant over short (t)

Axial (A) is dominant over terminal (a)

Purple flowers (P) is dominant over white (p)

Tall (T) is dominant over short (t)

Axial (A) is dominant over terminal (a)

Purple flowers (P) is dominant over white (p)

Write the phenotype for each.

Tt	tall
aa	terminal
PP	purple
TTpp	Tall AND white
AaPp	Axial AND purple

Tall (T) is dominant over short (t)

Axial (A) is dominant over terminal (a)

Purple flowers (P) is dominant over white (p)

Write the genotype for each.

Homozygous axial = AA

Short = tt

Heterozygous purple = Pp

Heterozygous tall AND terminal = Ttaa

Homozygous tall AND homozygous purple = TTPP

Heterozygous white = Not possible!

Mendel's 1st Law

- ***Monohybrid cross*** =
  - A cross of 2 individuals who are heterozygous for a single trait
  - Results in phenotypic ratio of 3:1
- ***Law of segregation*** =
  - Alleles of a gene are distributed into separate gametes during meiosis
  - AKA: Mendel's 1st Law

Monohybrid Cross

Parental generation (P_1)

Tall X Short

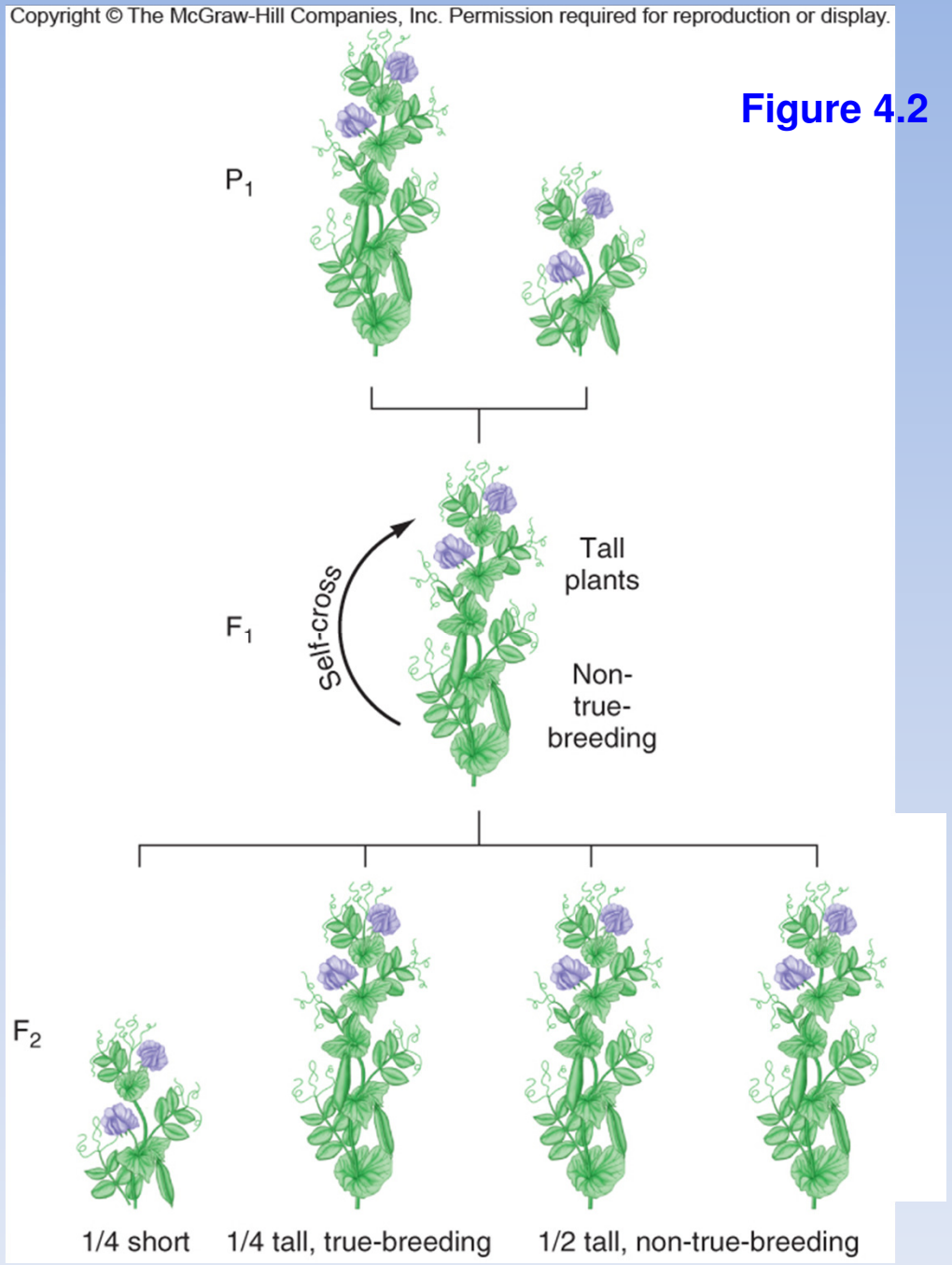
F_1

All Tall

F_2

1/4 Short : 3/4 Tall

Figure 4.2



Mendel's First Law – Segregation

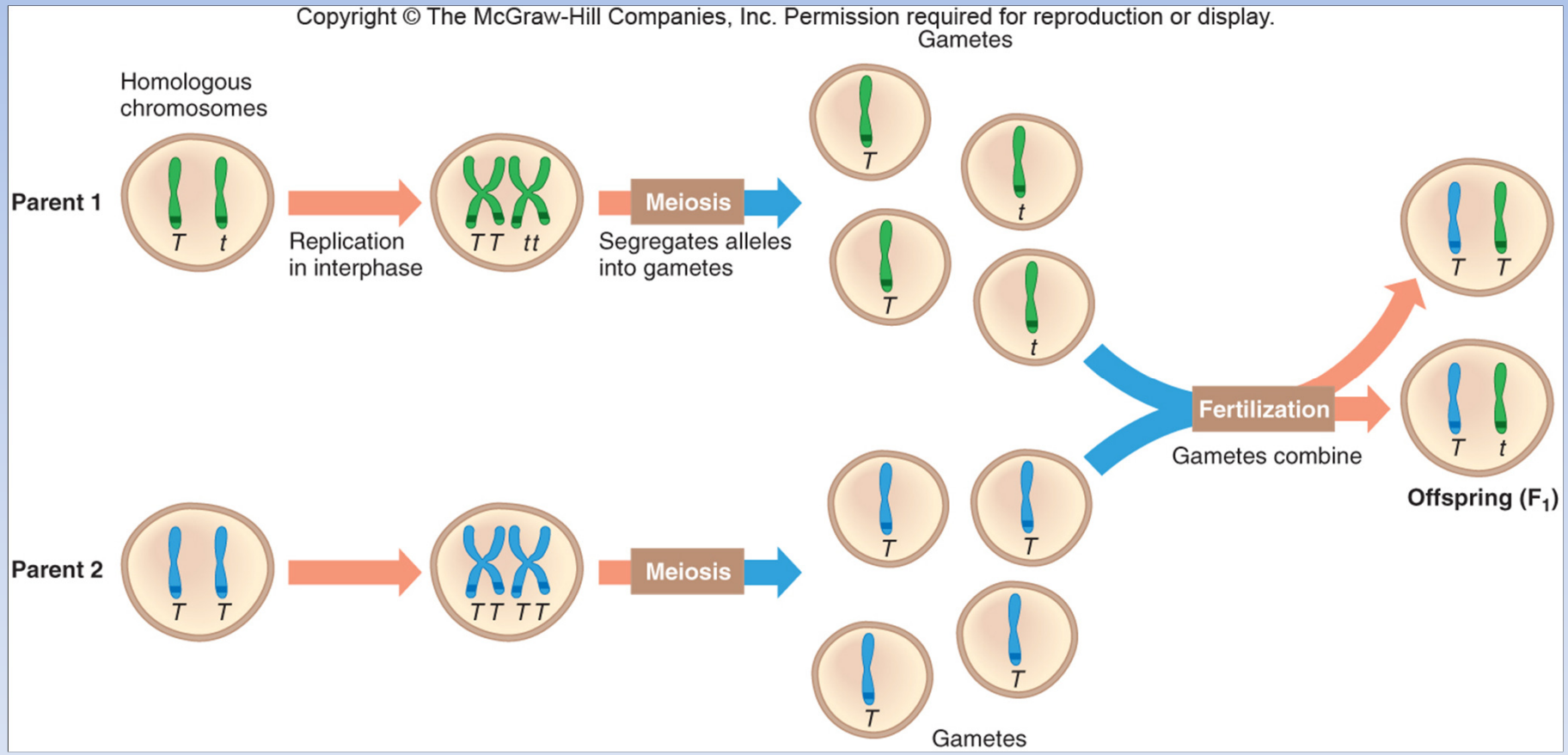


Figure 4.3

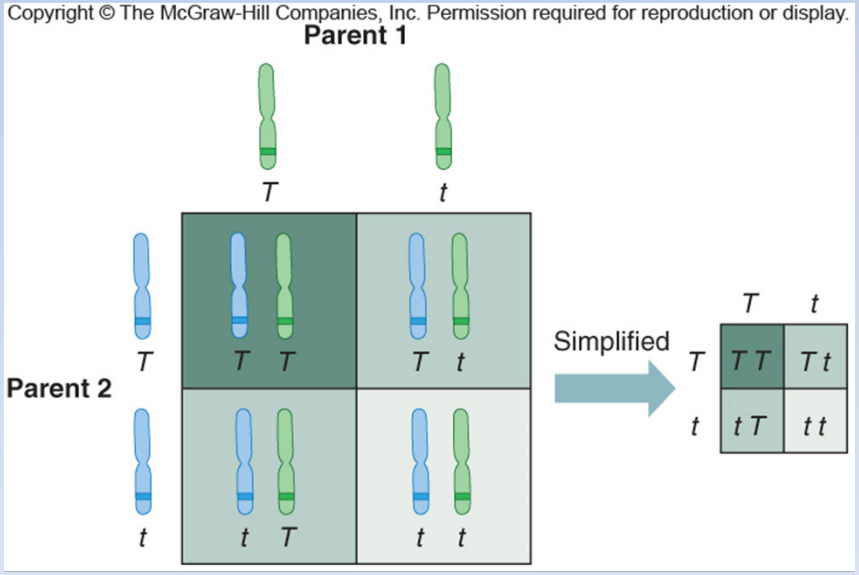
Practice Examples: 1 Trait

- ***Punnett Squares*** =
  - Represents particular genes in gametes and how they may combine in offspring

- Genotypic and

  Phenotypic Ratios
- Test Cross:

  Working Backwards



Modes of Inheritance

- ***Autosomal Dominant*** =
  - One autosomal allele causes a phenotype
  - Can affect males and females
  - Does not skip generations
  - Many diseases do not cause symptoms until adulthood
  - Example: Huntington disease

- ***Autosomal Recessive*** =
  - Two autosomal alleles are required to cause a phenotype
  - Can affect males and females
  - Can skip generations through carriers
  - Tend to be more severe and produce symptoms earlier
  - Example: Cystic fibrosis

Mendel's Second Law

- ***Law of independent assortment*** =
  - Inheritance of a gene on one chromosome does not influence inheritance of a gene on a different chromosome
  - AKA: Mendel's 2nd Law
- ***Dihybrid cross*** =
  - Breeding individuals that are heterozygous for 2 traits
  - Results in phenotypic ratio of 9:3:3:1

Mendel's Second Law – Independent Assortment

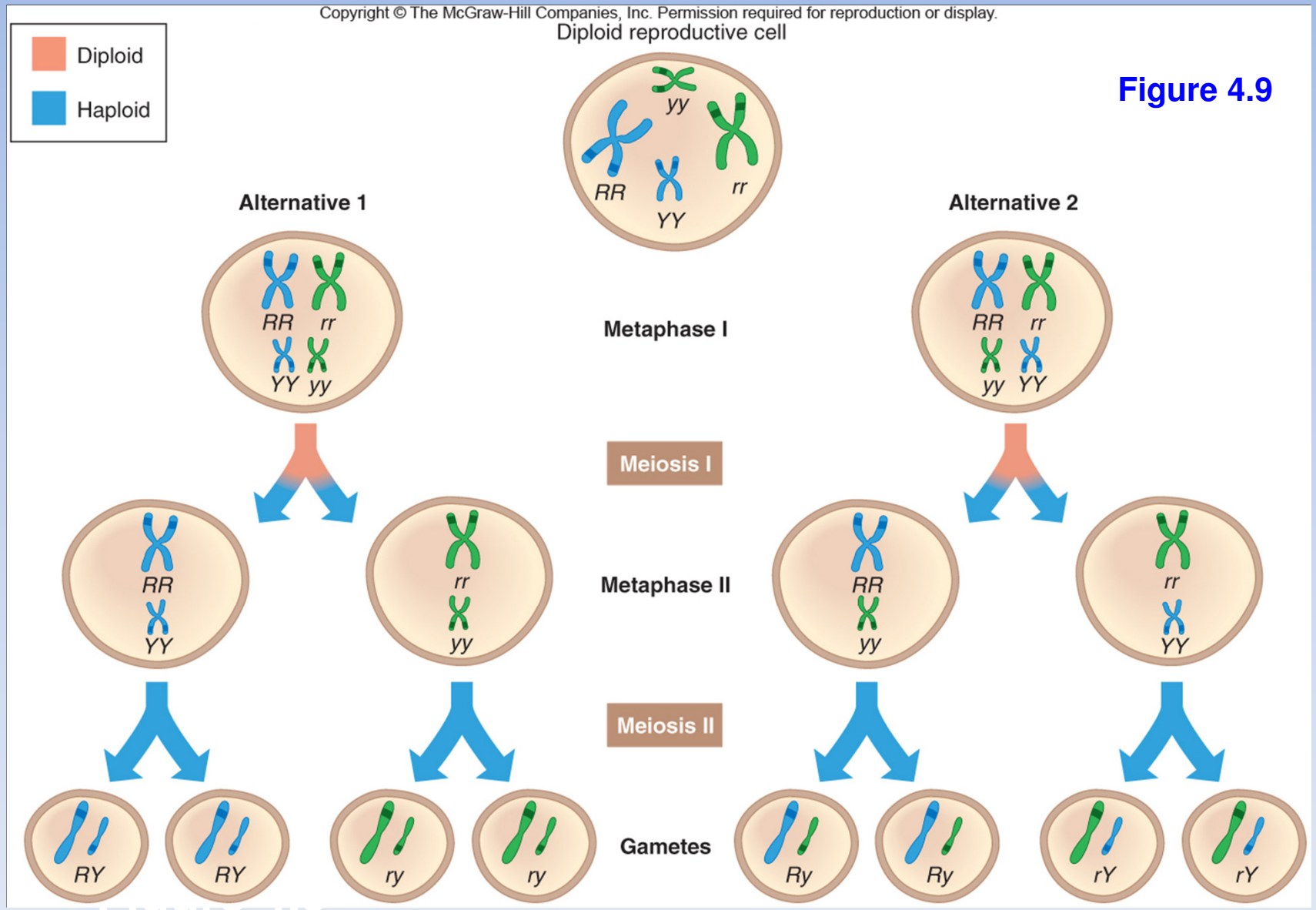


Figure 4.9

Practice Examples: 2 Traits

- Writing Genotypes and Phenotypes
- Punnett Squares
- Phenotypic Ratios

Mendel's Second Law – Independent Assortment

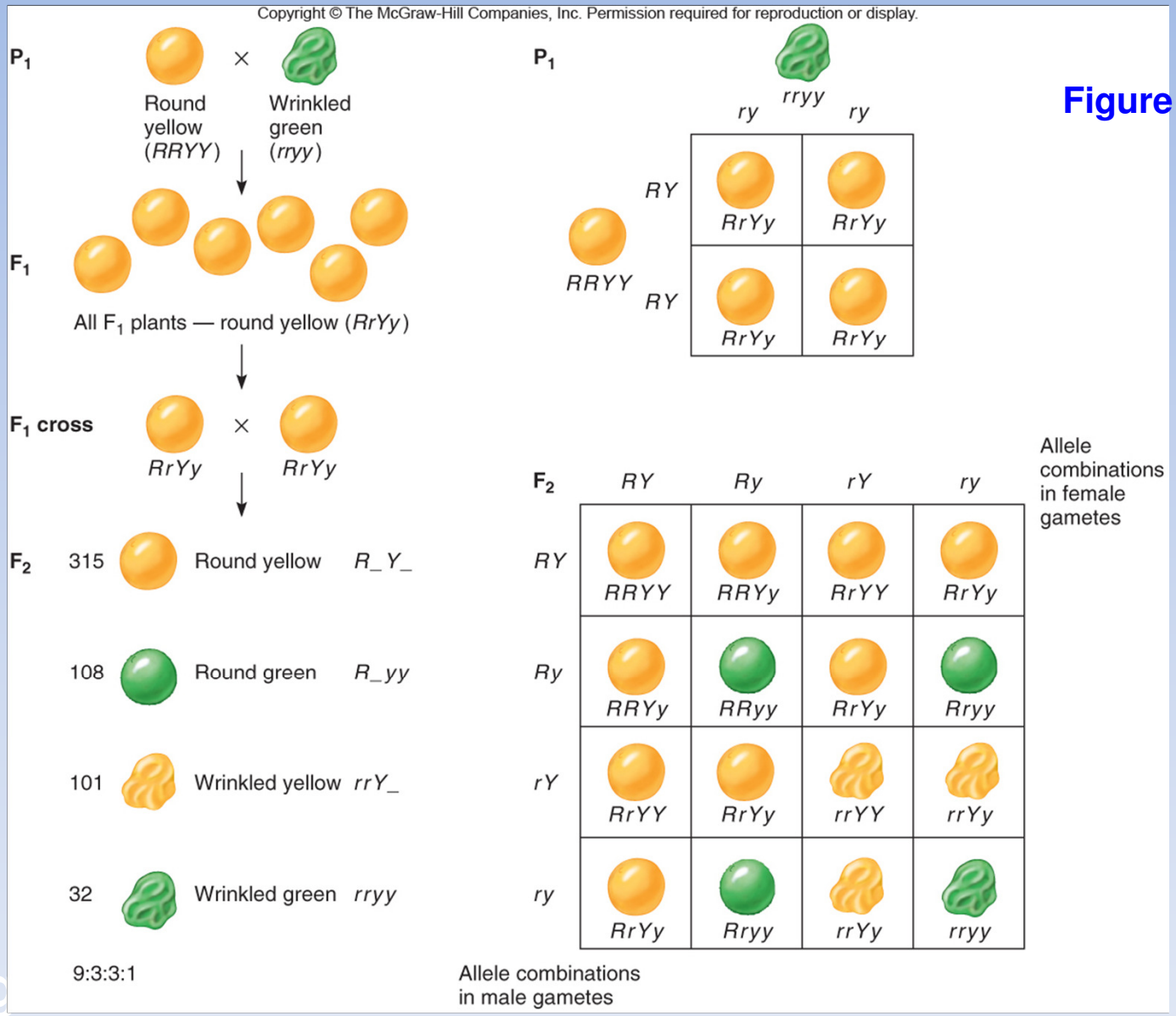


Figure 4.10

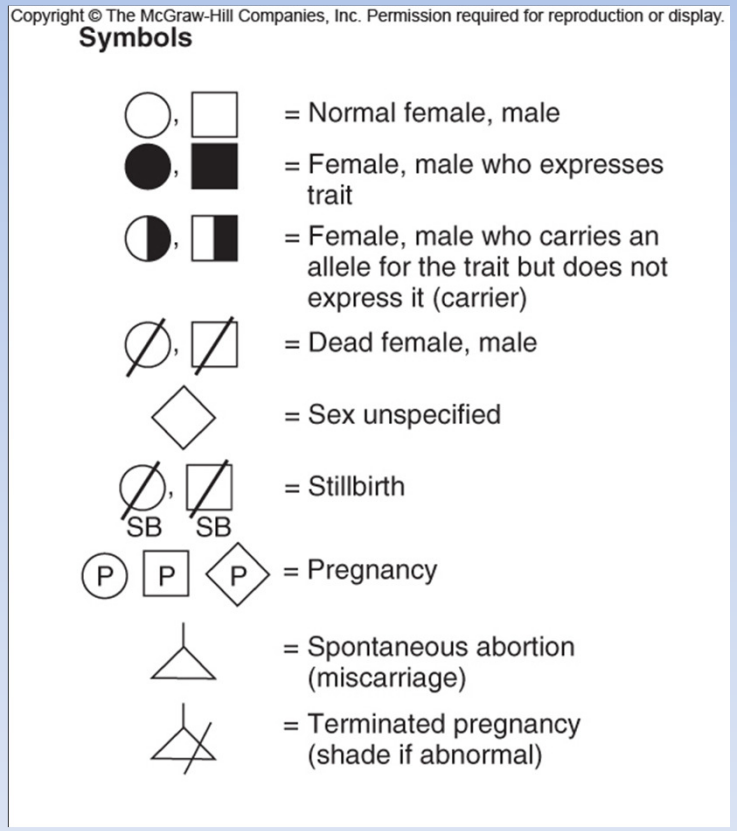
Pedigrees

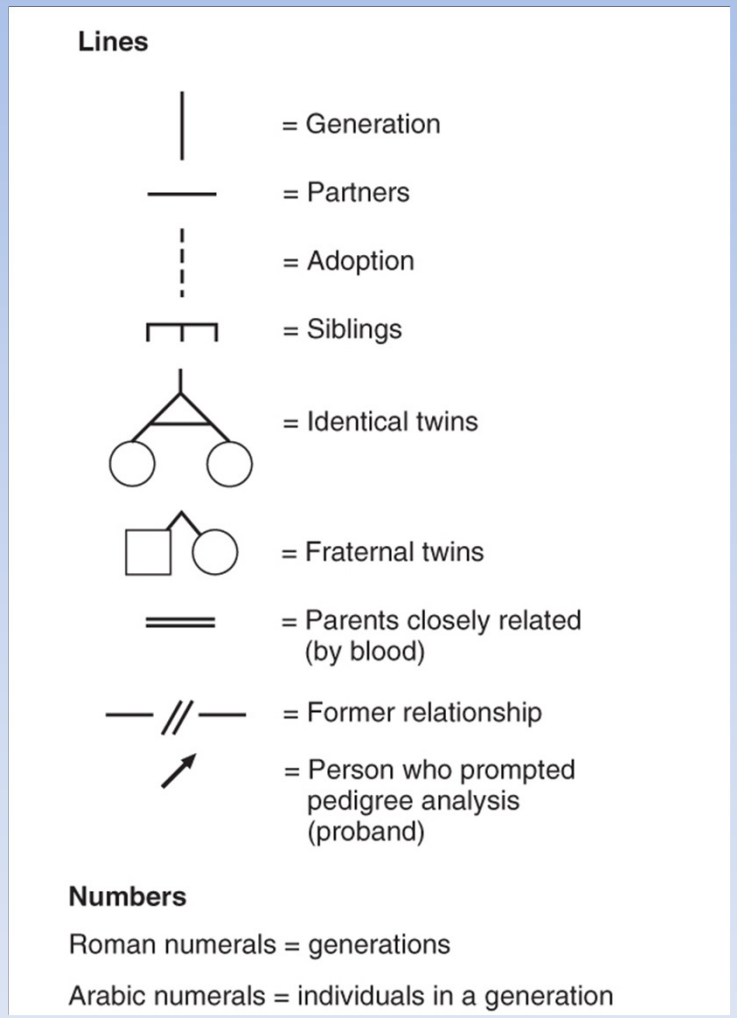
- ***Pedigree*** =
  - Chart of symbols connected by lines that depict the genetic relationships and transmission of inherited traits in related individuals

- Symbols
- Practice

Pedigree Analysis

Figure 4.13





Autosomal Recessive Trait

Albinism = Deficiency in melanin production

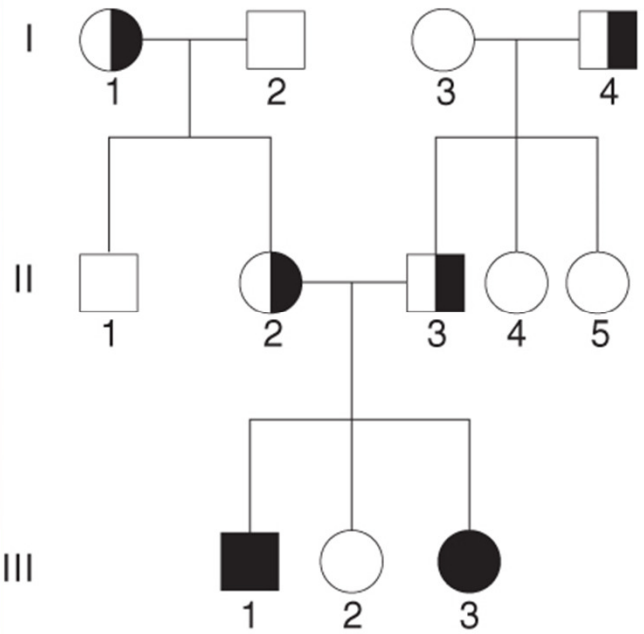
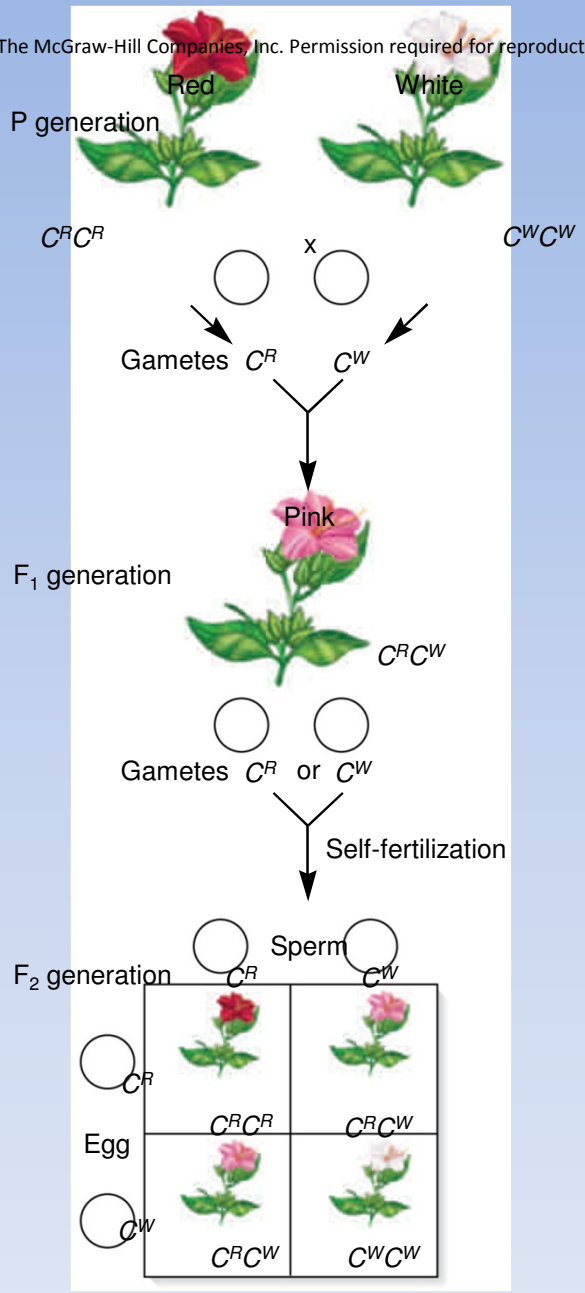


Figure 4.15

Beyond Mendel's Laws

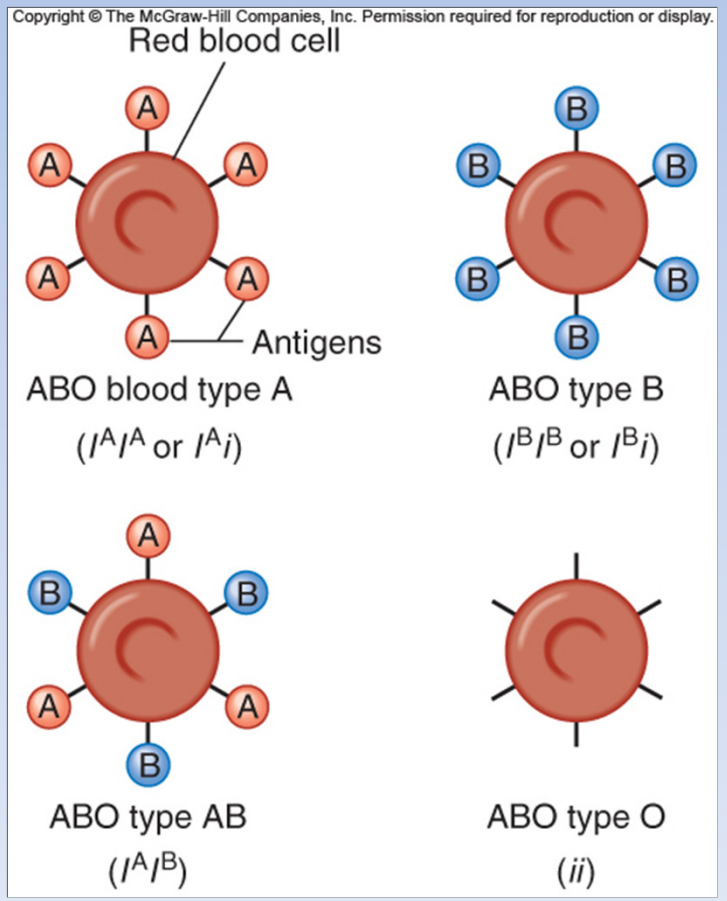
Incomplete Dominance

- The heterozygous phenotype is intermediate between that of either homozygous phenotype
- Example: Color of Four-O'Clock Plant
  - RR = Red
  - WW = White
  - RW = Pink



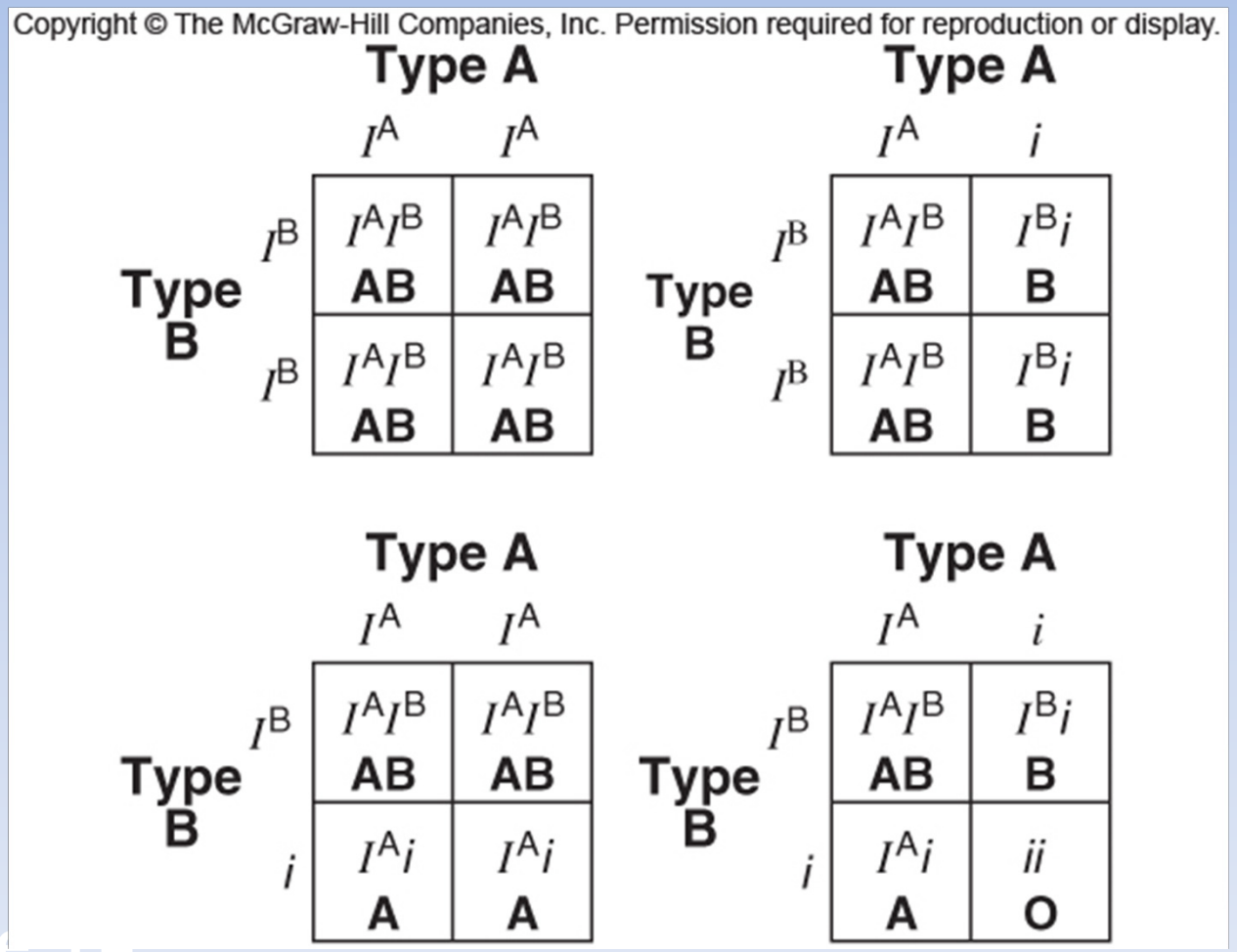
Codominance

- Heterozygote in which both alleles are fully expressed
- Example: Blood Type
  - I^AI^A or I^Ai= A
  - I^BI^B or I^Bi= B
  - I^AI^B = AB
  - ii = O



Offspring from Parents with Blood Type A and Blood Type B

Figure 5.4



Epistasis

- A gene masking the expression of another
- Refers to interaction between different genes, not between the alleles of the same gene
- Examples
  - Hairless gene in dogs
    - Genes that color hairs have no effect if there are no hairs
  - Albinism
    - One gene blocks the action of genes whose products confer color

Penetrance and Expressivity

- Degrees of expression of a single gene
  - Ex-polydactyly

- ***Penetrance*** =
  - Percentage of individuals with a genotype who have an associated phenotype
  - All-or-none expression of a genotype
- ***Expressivity*** =
  - Degree of severity of a phenotype

- ***Pleiotropy*** =
  - Single-gene disorder with several symptoms
    - Different symptom subsets may occur in different individuals
  - Ex-Marfan syndrome
- ***Genetic heterogeneity*** =
  - A phenotype that can be caused by variants (mutations) of any of several genes
  - Ex-Alzheimer disease
- ***Phenocopy*** =
  - An environmentally caused trait that occurs in a familial pattern, mimicking inheritance
  - Ex-infections like AIDS

Practice Examples: Incomplete Dominance and Codominance

- Writing Genotypes and Phenotypes
- Punnett Squares
- Genotypic and Phenotypic Ratios

Matters of Sex

Sex-Linked Traits

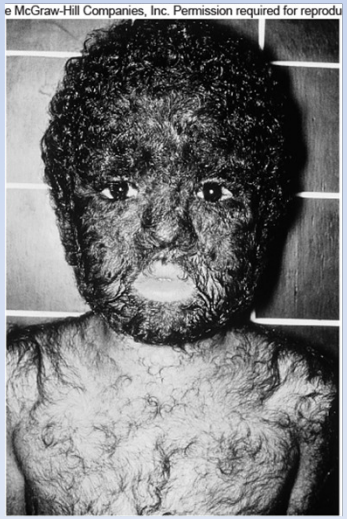
- Y-linked traits are passed on the Y chromosome
  - Rare because the Y chromosome has few genes
  - Passed from male to male
- X-linked traits are passed on the X chromosome
  - NO male to male transmission
  - Passed just like autosomal traits in females
    - Two copies required for expression of recessive allele
    - One copy for dominant
  - Males are ***hemizygous*** =
    - Have only 1 set of X-linked genes
    - Single copy causes expression of trait or illness

X-Linked Recessive Inheritance

- Males only need 1 recessive allele to express the trait
- Females need 2 copies of the recessive allele to express the trait
  - Considered a carrier if they inherit one copy
- Sons inherit trait from mother who is affected or is a carrier
- Daughters inherit trait from affected father AND mother who is affected or is a carrier
- Examples
  - Colorblindness
  - Hemophilia B

X-Linked Dominant Inheritance

- Expressed in females with at least one copy
- Much more severe in males
- High rates of miscarriage due to early lethality in males
- Passed from male to all daughters but to no sons
- Example
  - Congenital generalized hypertrichosis
    - Produces many extra hair follicles



Sex-Limited and Sex-Influenced Traits

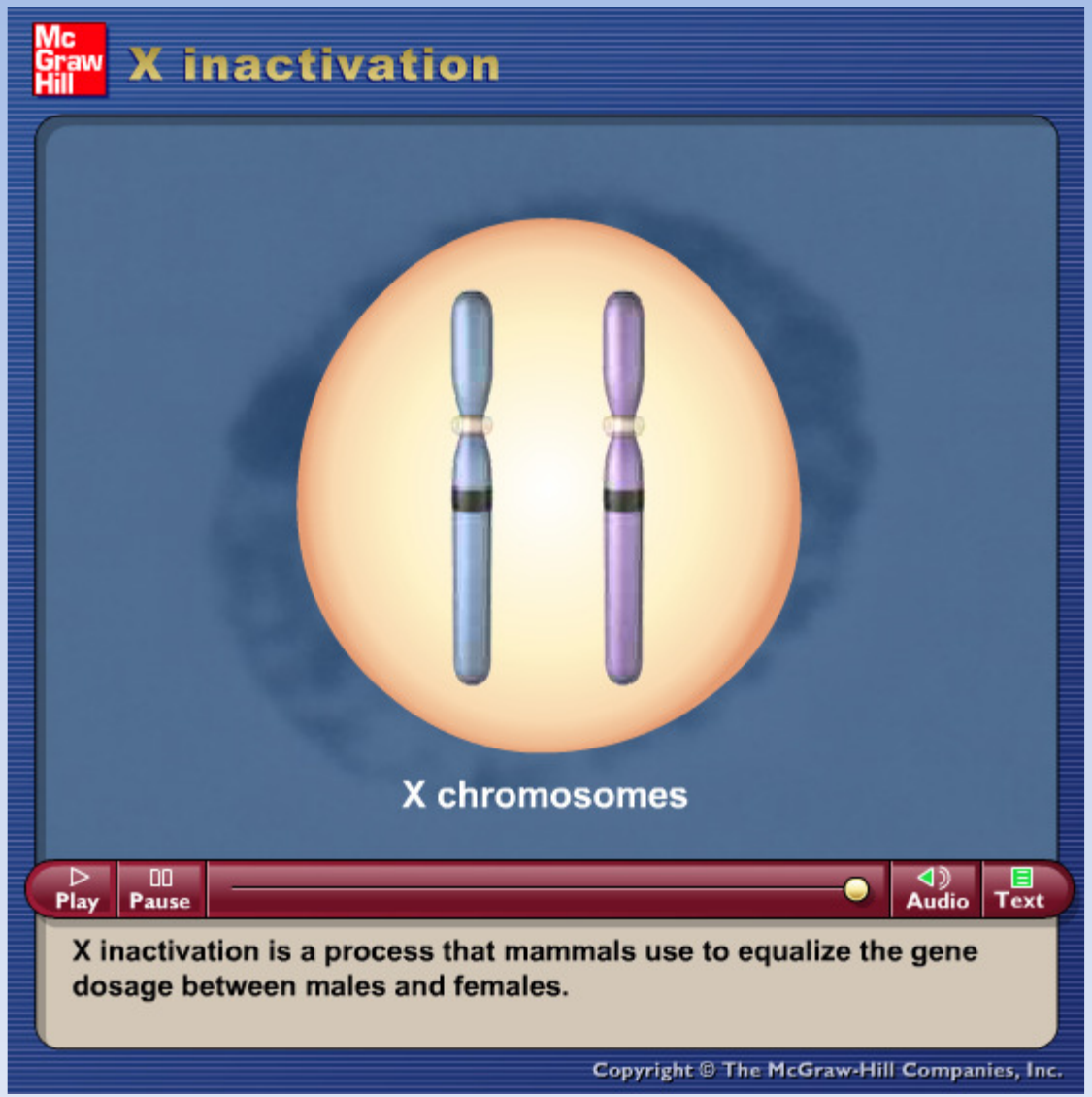
- ***Sex-limited trait*** =
  - Affects a structure or function present in only one sex
  - Examples
    - Beard growth
    - Preeclampsia (related to pregnancy)

- ***Sex-influenced trait*** =
  - Phenotype caused when an allele is recessive in one sex but dominant in the other
  - Example
    - Baldness

X Inactivation

- In female mammalian embryos
  - About 75% of the genes on one X chromosome in each cells are inactivated
  - Remaining 25% are expressed to different degrees in different women
  - “Mosaic”
- Inactive X chromosome is called a ***Barr body***
- ***Epigenetic*** =
  - Passed from one cell generation to the next but does not alter the DNA sequence

X Inactivation Animation

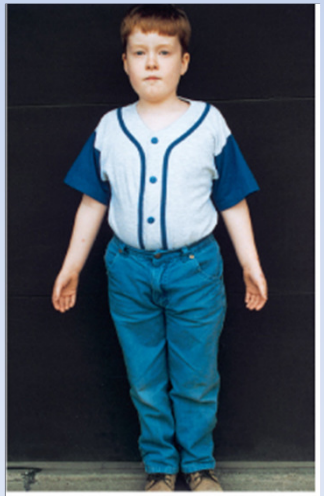


- ***<u>Manifesting heterozygote</u>*** =
  - Female carrier of an X-linked recessive gene who expresses the phenotype because the normal allele is inactivated in some tissues
  - Examples:
    - Calico cats
    - Rett syndrome

- X inactivation can be used to identify carriers for some X-linked disorders

Genomic Imprinting

- ***Genomic imprinting*** =
  - Differing of the phenotype depending upon which parent transmits a particular allele
  - Epigenetic
  - Imprinting pattern is passed from cell to cell in mitosis but not from individual to individual through meiosis
- Examples that result when nonimprinted copy of a gene is deleted
  - Prader-Willi Syndrome
  - Angelman Syndrome



b. Prader-Willi syndrome

c. Angelman syndrome
Angelman Syndrome Foundation

Practice Examples: X-Linked Inheritance

- Writing Genotypes and Phenotypes
- Punnett Squares
- Genotypic and Phenotypic Ratios
- Pedigrees